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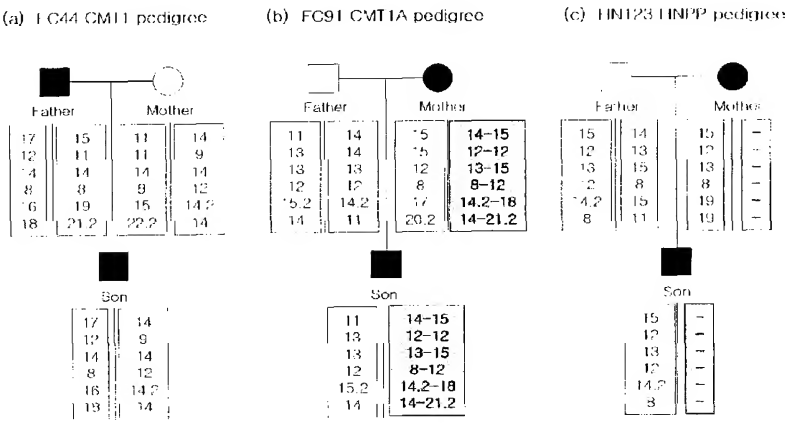
(54) Title: DIAGNOSIS METHOD AND KITS FOR INHERITED NEUROPATHIES CAUSED BY DUPLICATION OR DELETION OF CHROMOSOME 17P11.2-P12 REGION



(57) Abstract: Disclosed herein are a method and kit for diagnosing hereditary diseases CMT1A and HNPP, caused by duplicatino and deletion in the chromosome 17pl 1.2-pl2 region. In accordance with the present invention, there is provided a method for diagnosing an inherited neuropathy, comprising, runnung the PCR amplification using microsatellites present in a chromosome 17pl 1.2-pl2 region as markers and DNA typing the resulting PCR amplification products to determine the presence of duplication and deletion in the corresponding chromosomal region, wherein Multiplex PCR amplification is carried out using 6 loci of D17S921, D17S9B, D17S9A, D17S918, D17S2230 and D17S4A as markers, and DNA-typing of the resulting PCR amplification products is carried out to determine duplication and deletion in the corresponding chromosomal region. In accordance with the method of the present invention, the diagnosis accuracy of detecting duplication and deletion in the chromosome 17pl 1.2-pl2 region is greater than 99.9%.

WO 2006/011716 A1

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